

# [Breast cancer gene mutations biology essay](https://assignbuster.com/breast-cancer-gene-mutations-biology-essay-essay-samples/)

Breast Cancer affects nearly 1 in 4 women in the United States each year. Cancer is the uncontrolled growth of abnormal cells in the body. Breast cancer is a form of cancer that originates in the tissues of the breast. Using the latest technology researchers have determined that there is a specific line of genes linked to breast cancer, breast cancer (BRCA) 1 and BRCA 2 genes. The ability to test a patient for these breast cancer genes will help lead to better diagnosis and treatment for those with the gene.

Often breast cancer will start from a “ single abnormal cell that grows into a benign tumor” (Mader123). The tumor originates in the milk duct, where an extra lining of cells is formed that fills the ducts, this is known as ductal carcinoma. Another form, lobular carcinoma is when the cancer begins in the lobules, which are the glands that make the milk. Breast cancer is most common in females; however, males can still have breast cancer. In women it is recommended to be screened for breast cancer via self examination or mammograms, depending on age. Women who are over the age of forty are recommended to have a mammogram done every one to two years. Age is another risk because as a person gets older their risk increases. Race and ethnicity is another risk factor; white women are more at risk for breast cancer than African American women. Only “ about 5% to 10% of breast cancer cases are thought to be hereditary, resulting directly from gene defects inherited from a parent” (cancer. org).

Breast cancer can be classified by either BRCA 1 or BRCA 2 gene, which defines the mutation of the breast cancer gene. This form of breast cancer is considered genetic, which is when one dominate gene is passed onto subsequent generations and can result in the BRCA 1 or BRCA 2 gene. There are genes that can help with DNA repair, but in this case it is not a damaged gene it is mutated and cannot be repaired. Females who are heterozygous for the BRCA 1 mutation have a higher risk of getting breast cancer.

The gene loci for BRCA 1 gene can be found on the 27th chromosome, it is known as a tumor suppressor gene that provides the blueprint for protein that breaks cellular growth\*\*. Families who have history of breast and ovarian cancer have a strong occurrence of the BRCA1 gene and as a result there is a high risk for breast and ovarian cancer. Individuals that do have a family history of breast and ovarian cancers tend to acquire these cancers at a young age or acquire the more aggressive forms.

The gene loci for the BRCA 2 gene is found on the 13th chromosome; although breast cancer is rare in men, this gene tends to affect males at a higher rate. The gene for BRCA 2 is not expressed as often as BRCA 1, and families who carry this gene tend to be at a lower risk for developing cancer. Men who carry the BRCA 2 mutations are at a higher risk of developing breast cancer by age 70. Breast cancer is more commonly found in families with high male and female breast cancer occurrences. Breast cancer can also be related to ethnic background and specifically families who are of Jewish decent tend to be more likely to develop breast cancer due to a high mutation rate.

Prevalence of BRCA1 and BRCA2 mutations in different ethnic groups in the U. S.

## BRCA1

Asian American

0. 5%

African American

African American

1. 3-1. 4%

Caucasian

Caucasian (non-Ashkenazi Jewish)

2. 2-2. 9%

Hispanic

3. 5%

Ashkenazi Jewish

8. 3-10. 2%

Adapted from National Cancer Institute materials [5].

Figure : Prevalence of BRCA1 and BRCA2 mutations in different ethnic groups in the U. S. (komen. org) This table shows the percentages of the different ethnic groups and their chance of developing the BRCA1 or BRCA2 gene.

Men or women who are considered to be at high risk for breast cancer have the option to be tested. A person is considered to be at high risk if there are two or more family members that have had breast or ovarian cancer. The test for the breast cancer gene is a blood test that checks the DNA to see if there is a mutation in the BRCA 1 or BRCA 2 gene. A positive result of this test means that the patient has inherited the mutated gene. It is not definitive that the patient will get breast cancer as it requires unidentified environmental factors in order for the cancer to develop. However, a positive result means that the patient can pass the mutated gene to their children. If a person receives a negative result of having the breast cancer gene, it does not necessarily mean that they are not at risk for breast cancer, it means that they do not have the gene. In fact, “ 90 to 95 percent of breast cancer is not a result of BRCA 1 or BRCA 2, and 5 to 10 percent of women have the heredity factor of breast cancer” (Schnipper221). Also, “ The child of a parent who has a positive gene has a 50 percent risk of inheriting the mutation” (Schnipper223).

Testing for a genetic inheritance of the gene can be very helpful for families to know if they are carrying the mutated gene, and it can also be upsetting to the families who are affected. If a test comes back positive for the gene it will put other members of the family at risk for carrying the gene. Breast cancer heredity is very rare and it does not occur in every family. Age and ethnic background play a huge role in determining if you have a higher risk of carrying the gene. “ If a Jewish woman younger than 40 has breast cancer, there is about a 33% chance that they will be a carrier of the gene. While those who are not Jewish and have breast cancer before 30 have a 12% chance of having a mutation” (Love108). This illustrates that the genetic test is not a good determinant of risk for cancer. Positive results from the breast cancer test have a few options to lower their risk of getting breast cancer. Patients can choose to be monitored closer by their doctors for breast cancer signs by having frequent breast exams, mammograms, MRI’s, ultrasounds, blood tests, and also by self examination. A more aggressive step could be having prophylactic surgery, such as a double mastectomy. The surgery is a quite radical surgery and is not a common choice.

Research has indicated that those who carry either the BRCA1 or BRCA2 mutation can lower their risk of getting the cancer by keeping up with a physical, healthy lifestyle and by breastfeeding. Although that may not work on everyone, it has shown to help with some patients who carry the mutation. There is not much that can be done to prevent the chances of getting breast cancer besides radical surgery. There are other factors such as carrying the mutated breast cancer gene that can elevate the risk of getting breast cancer, a second primary cancer, which is a second breast tumor that is not related to the first tumor, can also show an increased risk of occurring with patients who carry the mutation. “ For BRCA1/2 carriers, the chance of a contralateral breast cancer 10 years after diagnosis of the first cancer is about 18 to 30 percent compared to about 10 percent for breast cancer survivors without a BRCA1/2 mutation” (komen. org).

Surgery, radiation therapy, tamoxifen, and chemotherapy are the most commonly used treatments for breast cancer. The treatment that is used depends on the type and severity of the cancer. A patient will go over the options that work best for them with their doctor to help get the best treatment. Two other surgeries that are performed depending on the patient’s cancer are Lumpectomy, and Mastectomy. Lumpectomy is a surgery that removes only the cancerous lump from the breast. Mastectomy is removal of the entire breasts to remove the cancer and the tissue that surrounds it. Men and women who have breast cancer as a result of a genetic mutation will go through the same treatments. A patient who has a genetic mutation of breast cancer does not have their own treatment plan. Although they can take precautions to help prevent their chance of developing breast cancer, there is no different treatment if they do have breast cancer.

“ Only five to ten percent of breast cancer patients in the United States are linked to a high risk genetic mutation” (komen. org). Men and women who inherited the BRCA 1 or BRCA 2 gene, do not make up the population of those who suffer from breast cancer. Having a genetic mutation is not the only way for a patient to receive breast cancer, there are many other aspects. There are chances that getting breast cancer may be genetically linked, but there is still research being conducted to investigate the occurrence of breast cancer.